

HB 6580

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### TESTIMONY TO ADVOCATE FOR AN ACT TO CREATE CONNECTICUT RARE DISEASE ADVOSORY COUNCIL

In the state of Connecticut with its population of 3.6 million, it is estimated that 720 – 3600 individuals suffer from rare diseases. Majority of these are inherited disease but some do not have clear-cut genetic inheritance. Generally these are complex disorders with large unmet treatment need and poor access to healthcare. There are few doctors who have expertise or interest in care of these individuals. Thus many patients may go without a correct diagnosis from physician to physician often for years developing disabling complications. Even after the diagnosis is made, patients do not have the usual supports such as social work, psychological expertise, physical therapy or financial coordinators. These types of support are readily available and expected as standard of care for other disease, for example patients with liver disease due to hepatitis C or alcohol who require liver transplantation.

Patients and families affected by rare disease face a life-long battle with their disease and often have to educate themselves more than physicians due to lack of access to expert care. These patients and families deserve better from our health care system. When patients with rare disease are provided optimal care, they experience marked improvement in their quality of life and functional status. Experience with a prototype rare disease, Gaucher disease have shown that access to optimal care result in high-functioning individuals who make important contributions to society at large.

I therefore, most emphatically support the act to establish an advisory council to explore the issue of rare diseases and make recommendations for improving awareness, funding and assistance to persons suffering from rare diseases in Connecticut.

